hand-foot-genital syndrome

Hand-foot-genital syndrome is a rare condition that affects the development of the hands and feet, the urinary tract, and the reproductive system. People with this condition have abnormally short thumbs and first (big) toes, small fifth fingers that curve inward (clinodactyly), short feet, and fusion or delayed hardening of bones in the wrists and ankles. The other bones in the arms and legs are normal.

Abnormalities of the genitals and urinary tract can vary among affected individuals. Many people with hand-foot-genital syndrome have defects in the ureters, which are tubes that carry urine from each kidney to the bladder, or in the urethra, which carries urine from the bladder to the outside of the body. Recurrent urinary tract infections and an inability to control the flow of urine (urinary incontinence) have been reported. About half of males with this disorder have the urethra opening on the underside of the penis (hypospadias).

People with hand-foot-genital syndrome are usually able to have children (fertile). In some affected females, problems in the early development of the uterus can later increase the risk of pregnancy loss, premature labor, and stillbirth.

Frequency

Hand-foot-genital syndrome is very rare; only a few families with the condition have been reported worldwide.

Genetic Changes

Mutations in the *HOXA13* gene cause hand-foot-genital syndrome. The *HOXA13* gene provides instructions for producing a protein that plays an important role in development before birth. Specifically, this protein appears to be critical for the formation and development of the limbs (particularly the hands and feet), urinary tract, and reproductive system. Mutations in the *HOXA13* gene cause the characteristic features of hand-foot-genital syndrome by disrupting the early development of these structures. Some mutations in the *HOXA13* gene result in the production of a nonfunctional version of the HOXA13 protein. Other mutations alter the protein's structure and interfere with its normal function within cells. Mutations that result in an altered but functional HOXA13 protein may cause more severe signs and symptoms than mutations that lead to a nonfunctional HOXA13 protein.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Hand-foot-uterus syndrome
- HFG syndrome
- HFGS
- HFU syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hand foot uterus syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1841679/

Other Diagnosis and Management Resources

- GeneReview: Hand-Foot-Genital Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1423
- MedlinePlus Encyclopedia: Hypospadias
  https://medlineplus.gov/ency/article/001286.htm
- MedlinePlus Encyclopedia: Urinary Tract Infection
  https://medlineplus.gov/ency/article/000521.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Hypospadias
  https://medlineplus.gov/ency/article/001286.htm
- Encyclopedia: Urinary Tract Infection
  https://medlineplus.gov/ency/article/000521.htm

- Health Topic: Foot Injuries and Disorders
  https://medlineplus.gov/footinjuriesanddisorders.html
- Health Topic: Hand Injuries and Disorders
  https://medlineplus.gov/handinjuriesanddisorders.html

Genetic and Rare Diseases Information Center

- Hand foot uterus syndrome
  https://rarediseases.info.nih.gov/diseases/2594/hand-foot-uterus-syndrome

Educational Resources

- American Society for Surgery of the Hand: Congenital Hand Differences
  http://www.assh.org/handcare/hand-arm-conditions/Congenital-Differences
- Cleveland Clinic: Congenital Hand Differences
  http://my.clevelandclinic.org/health/articles/congenital-hand
- Disease InfoSearch: Hand Foot Uterus Syndrome
  http://www.diseaseinfosearch.org/Hand+Foot+Uterus+Syndrome/3232
- MalaCards: hand-foot-uterus syndrome
  http://www.malacards.org/card/hand_foot_uterus_syndrome
- Orphanet: Hand-foot-genital syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2438

Patient Support and Advocacy Resources

- March of Dimes: Genital and Urinary Tract Defects
  http://www.marchofdimes.org/baby/genital-and-urinary-tract-defects.aspx
- Resource list from the University of Kansas Medical Center: Limb Anomalies
  http://www.kumc.edu/gec/support/limb.html

GeneReviews

- Hand-Foot-Genital Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1423

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28hand-foot-genital+syndrome%29+OR+%28hand-foot-uterus+syndrome%29%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- HAND-FOOT-GENITAL SYNDROME
  http://omim.org/entry/140000

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